

Positive Result:

Blood Spot Screen Result Notification



Elevated C3-DC Acylcarnitine

Next Steps

This week, you should take the following recommended actions:

- **Consult** with a metabolic specialist. Contact information for the metabolic specialists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Evaluate** infant for signs of hypoglycemia or ketonuria; arrange immediate referral if symptomatic.
- **Arrange** referral to a metabolic specialist for further diagnostic work-up.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share the follow-up plan with them. Educate family about signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by specimen collection before 24 hours.

Differential Diagnosis

Elevated C3-DC acylcarnitine is primarily associated with:

- Malonic acidemia — Incidence is unknown

Clinical Summary

Malonic acidemia is an organic acid disorder. People with malonic acidemia are unable to break down fatty acids for energy, which results in an increase in malonic acid and its derivatives. Infants may present acutely with hypoglycemia, lactic acidosis, and severe lethargy. Almost all affected children have delayed development. If untreated, symptoms can progress to metabolic acidosis, vomiting, failure to thrive, hypotonia, cardiomyopathy, and seizures.

Treatment consists of a high-fat restricted diet and avoidance of fasting. Supplements may also be prescribed.

Because of how rare malonic acidemia is, the expected outcomes following treatment is not well known. Without treatment, children with malonic acidemia could die in infancy.